Peutz–Jeghers syndrome: A very rare cause of iron deficiency anemia

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A 11-year-old boy was admitted to clinic with a one year history of fatigue and abdominal pain. In physical examination, there were multiple brown-violet colored, small flat pigmentations on his buccal mucosa, lips and brown spotty pigmentations on the lip mucosa from his birth (Figure 1). His mother also had similar mucocutaneous pigmentation and was operated for intestinal polyps (Figure 2).

Laboratory findings were consistent with severe iron deficiency anemia and fecal occult blood test was positive. Gastrointestinal endoscopic examination revealed two polyps at the stomach and three polyps at jejunum that caused bleeding were removed with forceps (Figure 3).

Histopathologic examination revealed hamartomatous polyps. The presence of brown pigmentations and multiple gastrointestinal polyps alerted us to a possible diagnosis of Peutz Jeghers syndrome and serine/threonine kinase 11 (STK11, also named LKB1) mutation was found positive both the patient and his mother.

It is very important to conduct a thorough physical examination and to probe the family history in the cases of iron deficiency anemia that is frequently encountered in children, especially in the presence
of other complaints such as abdominal pain (1,2). This allows early diagnosis of rare diseases such as PJS, which leads to high risk of developing cancer, and examination family members for the associated complications by using advanced diagnostic tools (3,4,5).

References


